

Figure 1A

ESTs cluster 1  
ESTs cluster 2  
ESTs cluster 3

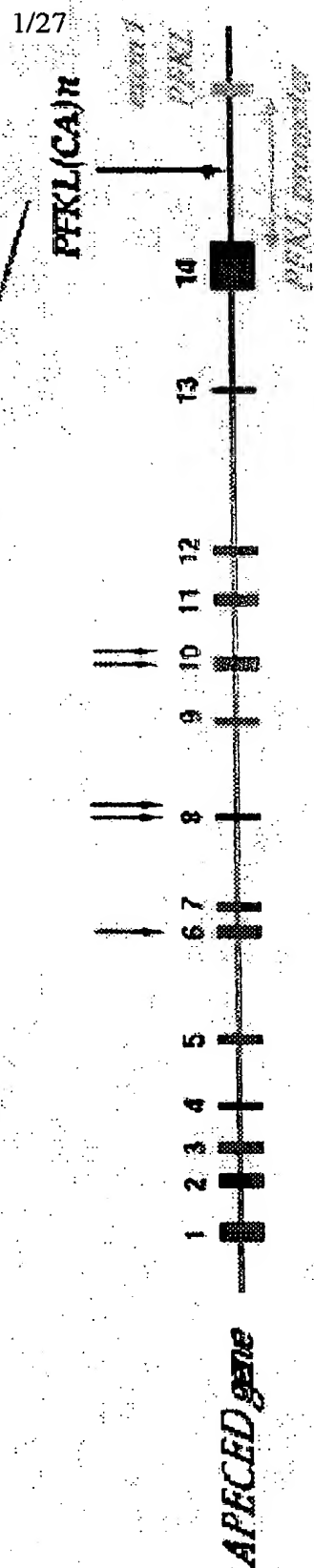


Figure 1B

Exon predictions  
for APECED

Grail2	Grail3	Gene	Xpound
1	1	1	1
2	2	2	2
3	3	3	3
4	4	4	4
5	5	5	5
6	6	6	6
7	7	7	7
8	8	8	8
9	9	9	9
10	10	10	10
11	11	11	11
12	12	12	12
13	13	13	13
14	14	14	14
15	15	15	15
16	16	16	16
17	17	17	17
18	18	18	18
19	19	19	19
20	20	20	20
21	21	21	21
22	22	22	22
23	23	23	23
24	24	24	24
25	25	25	25
26	26	26	26
27	27	27	27

005020" 56560560

cDNA B1-1

Q21D1

Q11D11



Figure 1C

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1 cgggagcagccggcgaggagggcccccacagcccccgccgggagccccgagggccaagcgagg 60  
 61 gctgccagtgtcccgagccaccacccgctccgccccccagcccccggtccccggcccccccc 120  
 121 atggcgacggagcgcgctacgcccgttcttgaggctgcaccgcacgagagatcgcggtg 180  
 1 M A T D A A L R R L L R L H R T E I A V 20  
 181 gccgtgacagcgcccttcccactgtgcacgctggtgctgaccacgacgtggtccccgag 240  
 21 A V D S A F P L L H A L A D H D V V P E 40  
 241 gacaagtttcaggagacgcttcatctgaaggaaaaggaggtgctgccccagggccttccac 300  
 41 D K F Q E T L H L K E K E G C P Q A F H 60  
 301 gccctcctgtcctggctgacctgacctccacagccatcctggacttctggagggtg 360  
 61 A L L S W L L T Q D S T A I L D F W R V 80  
 361 ctgttcaaggactacaacctggagcgctatggccggctgcagccccatcctggacagcttc 420  
 81 L F K D Y N L E R Y G R L Q P I L D S F 100  
 421 cccaaagatgtggacctcagccagccccggaaaggaggaggaagcccccgccgtccccaaag 480  
 101 P K D V D L S Q P R K G R K P P A V P K 120  
 481 gctttgtaccgcccaccagactccccaccacgaaggaaagccctcagaagaggctcagct 540  
 121 A L V P P P R L P T K R K A S E E A R A 140  
 541 gccgagcagcagccctgactccaaggggcaccgcccagccaggtctctcaactgaaggcc 600  
 141 A A P A A L T P R G T A S P G S Q L K A 160  
 601 aagcccccaagaagccggagagcagcgagcagcagcgcccttccactcgggaacggg 660  
 161 K P P K K P E S S A E Q Q R L P L G N G 180  
 661 attcagaccatgtcagcttccagtcagagagctgtggccatgtcctcctccgggacgtcccg 720  
 181 I Q T M S A S V Q R A V A M S S G D V P 200

FIGURE 2A

721	ggagcccgaggggcccgtggaggggacccatccatccagcaggtgttgcagtcaggcggtccc	780
201	G A R G A V E G I L I Q Q V F E S G G S	220
781	aagaagtgcacccaggttggtggggagttctacactccagcaagttcgaagactccggc	840
221	K K C I Q V G G E F Y T P S K F E D S G	240
841	agtggaagaacaaggccgcagcagcagtcagtgggcccgaaagcctctgttcgagccaaggga	900
241	S G K N K A R S S S G P K P L V R A K G	260
901	gccagggcgctgccccgggtggaggtgaggtgaggtggtggccagcagggcagcgttccc	960
261	A Q G A A P G G E A R L G Q Q G S V P	280
961	gccccctctgccccctccagtgacccccagctccaccagaagaatgaggacgagtggtgcc	1020
281	A P L A L P S D P Q L H Q K N E D E C A	300
1021	gtgtgtcgggacggcggtgagctcatctgtgtgacggctgccccctgggccttccacctg	1080
301	V C R D G G E L I C C D G C P R A F H L	320
1081	gcctgcctgtccccctccgctccgggagatccccagtcgggacctggagggtgctccagctgc	1140
321	A C L S P P L R E I P S G T W R C S S C	340
1141	ctgcaggcaacagtcacaggaggtgcagccccgggcagaggagccccggccccagggagcca	1200
341	L Q A T V Q E V Q P R A E E P R P Q E P	360
1201	cccgtggagacccccgctccccccggggttaggtcgccgggagaggaggttaagaggtcca	1260
361	P V E T P L P P G L R S A G E E V R G P	380
1261	cctgggggaacccctagccggcatggacacgactctgtctacaagcacctgcccggctccg	1320
381	P G E P L A G M D T T L V Y K H L P A P	400
1321	ccttctgcagccccgctgccagggctgggactcctcgccctgcacccccctactgtgtgtg	1380
401	P S A A P L P G L D S S A L H P L L C V	420

1381 ggtcctgagggtcagcagaacctggctcctgggtgcgcgttgccgggtgtgcggagatggt 1440  
 421 G P E G Q Q N L A P G A R C G V C G D G 440  
 1441 acggacgtgctgcggtgtactcactgcgcgctgccttccactggcgtgcccacttccca 1500  
 441 T D V L R C T H C A A A F H W R C H F P 460  
 1501 gccggcacctcccgcccgaggacgggacctgcgtgcagatcctgctcaggagacgtgacc 1560  
 461 A G T S R P G T G L R C R S C S G D V T 480  
 1561 ccagccctgtggagggtgtgctggccccagccccgcctggccccctggccctgcc 1620  
 481 P A P V E G V L A P S P A R L A P G P A 500  
 1621 aaggatgacactgccagtcacgagcccgctctgcacagggatgacctggagtccttctg 1680  
 501 K D D T A S H E P A L H R D D L E S L L 520  
 1681 agcagcacacaccttcgatggcatcctgcagtgggccatccagagcatggccccctccggcg 1740  
 521 S E H T F D G I L Q W A I Q S M A R P A 540  
 1741 gcccccttccctcctgacccccagatggccgggacatgcagctctgatgagagtgctg 1800  
 541 A P F P S 546  
 1801 agaaggacacctcctcctcagtcctggaagccggccggtggatcaagaagggacag 1860  
 1861 cgccacctctgtcagtgctcggtgtaaacagctctgtgtttcttggggacaccagccat 1920  
 1921 catgtgcctggaaattaaacctgccccacttcttactctggaagtcctccgggagcctc 1980  
 1981 tccttgctggtgacctaataataaaaaattagctgggtgtggtgggtgcctg 2040  
 2041 taatcccagctacatgggagcctgaggcatgagaatcacttgaactcgggaggtgaggt 2100  
 2101 tgcagtgagctgagattggccactgcactccagctgtggtcggaagagtgcactccgt 2160  
 2161 ctcaaaaaacaaaaaa<sup>pl-1</sup>accacataacataaatttatcatctcgaccacttttcagt 2220  
 2221 tcagtggcattcacatctcatgtaa 2245

FIGURE 2A (cont.')

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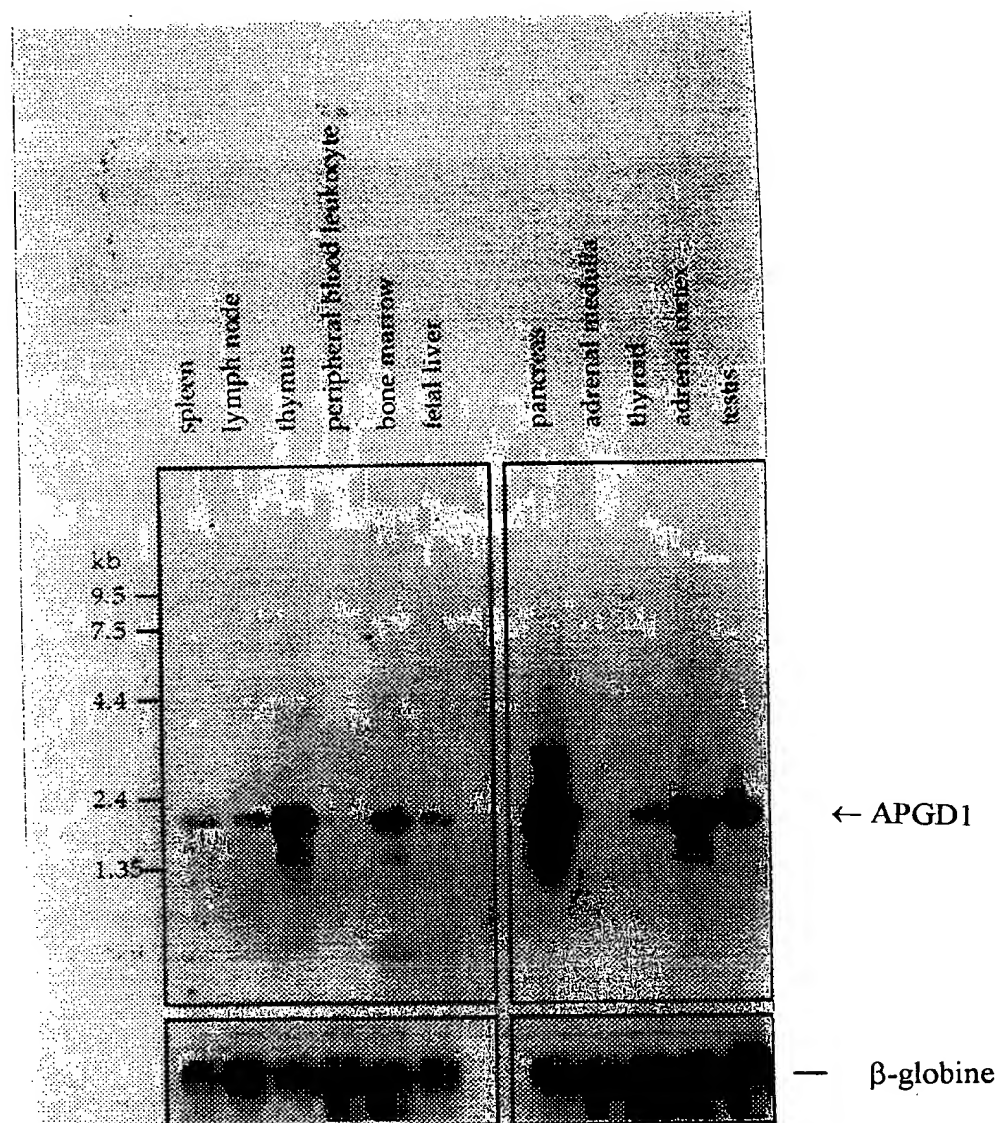
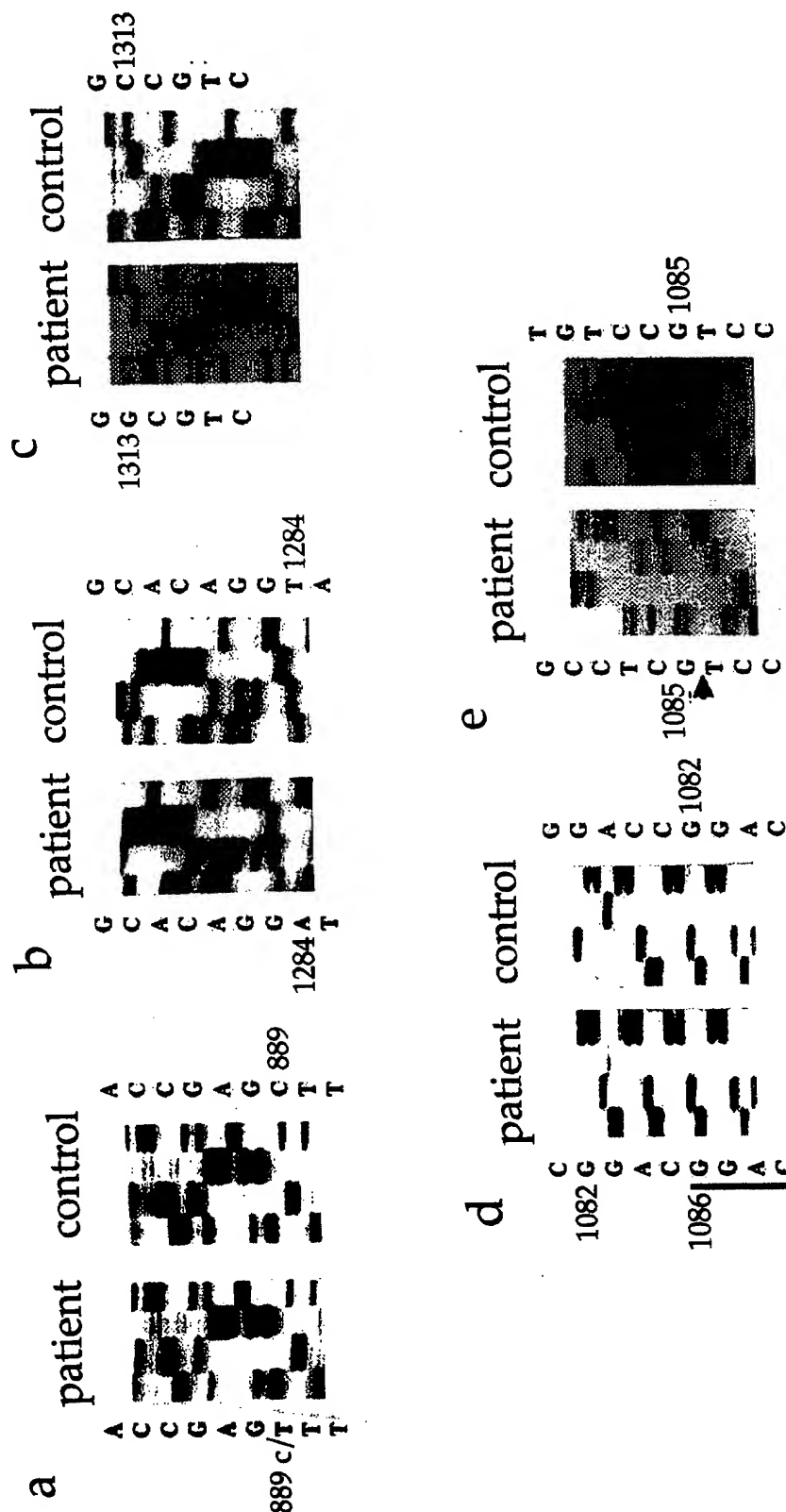


Figure 2B



The sequence lanes appear from left to right, as C, A, T, and G

Figure 3

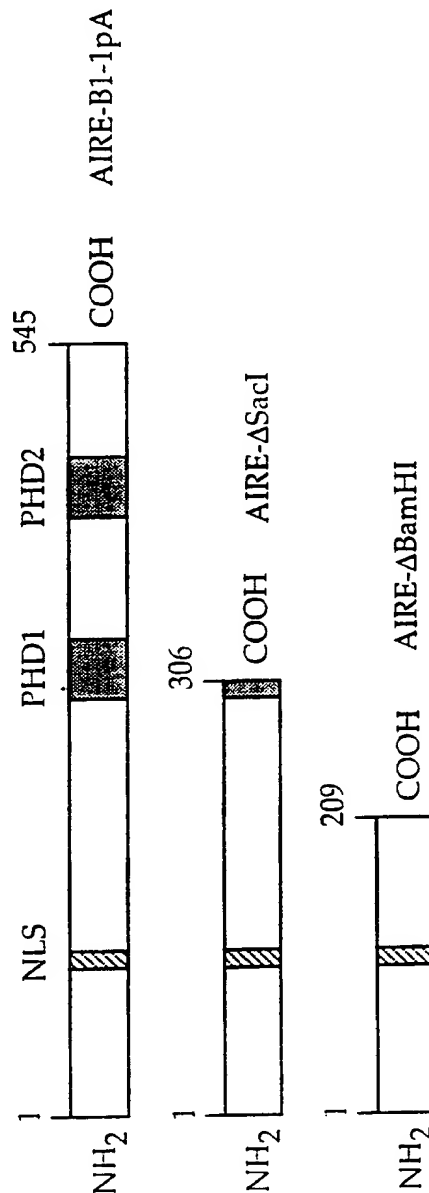


Figure 4



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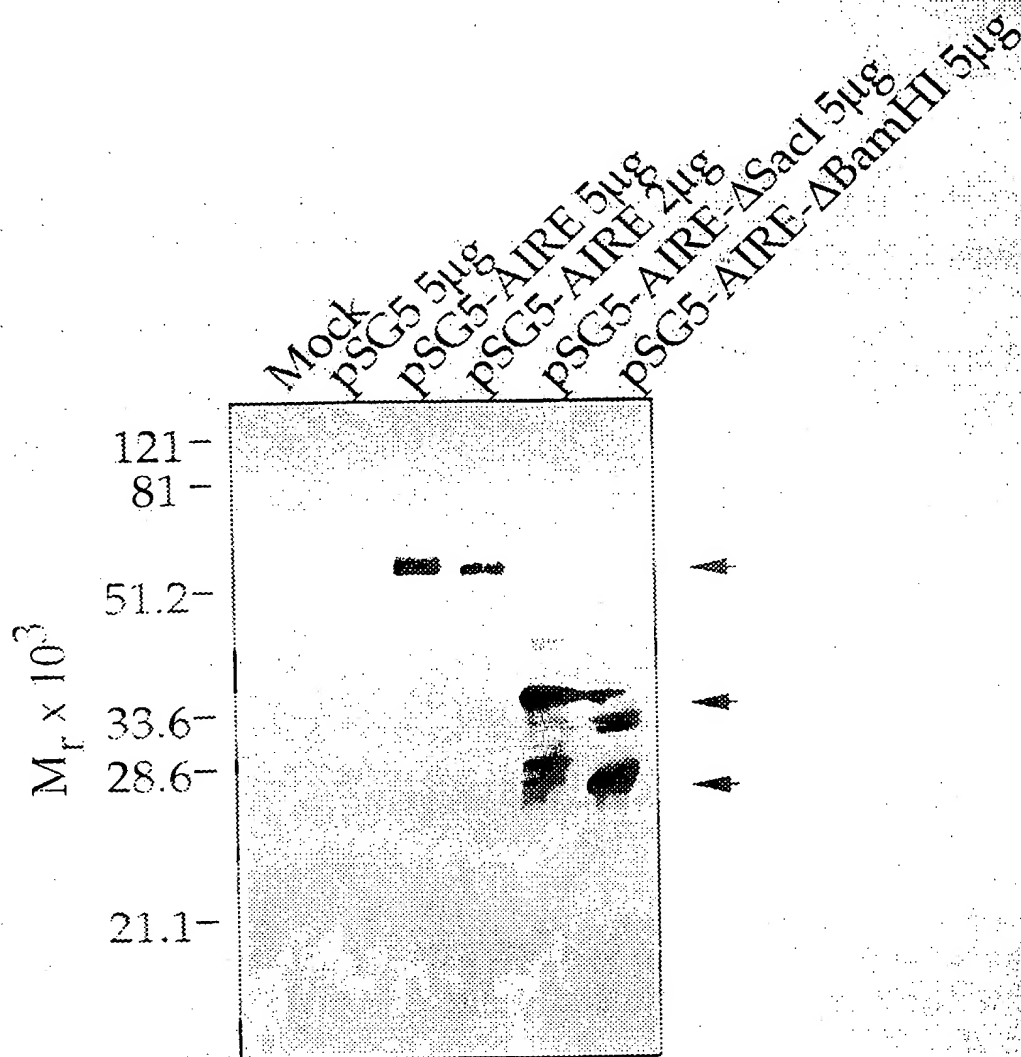


Figure 5

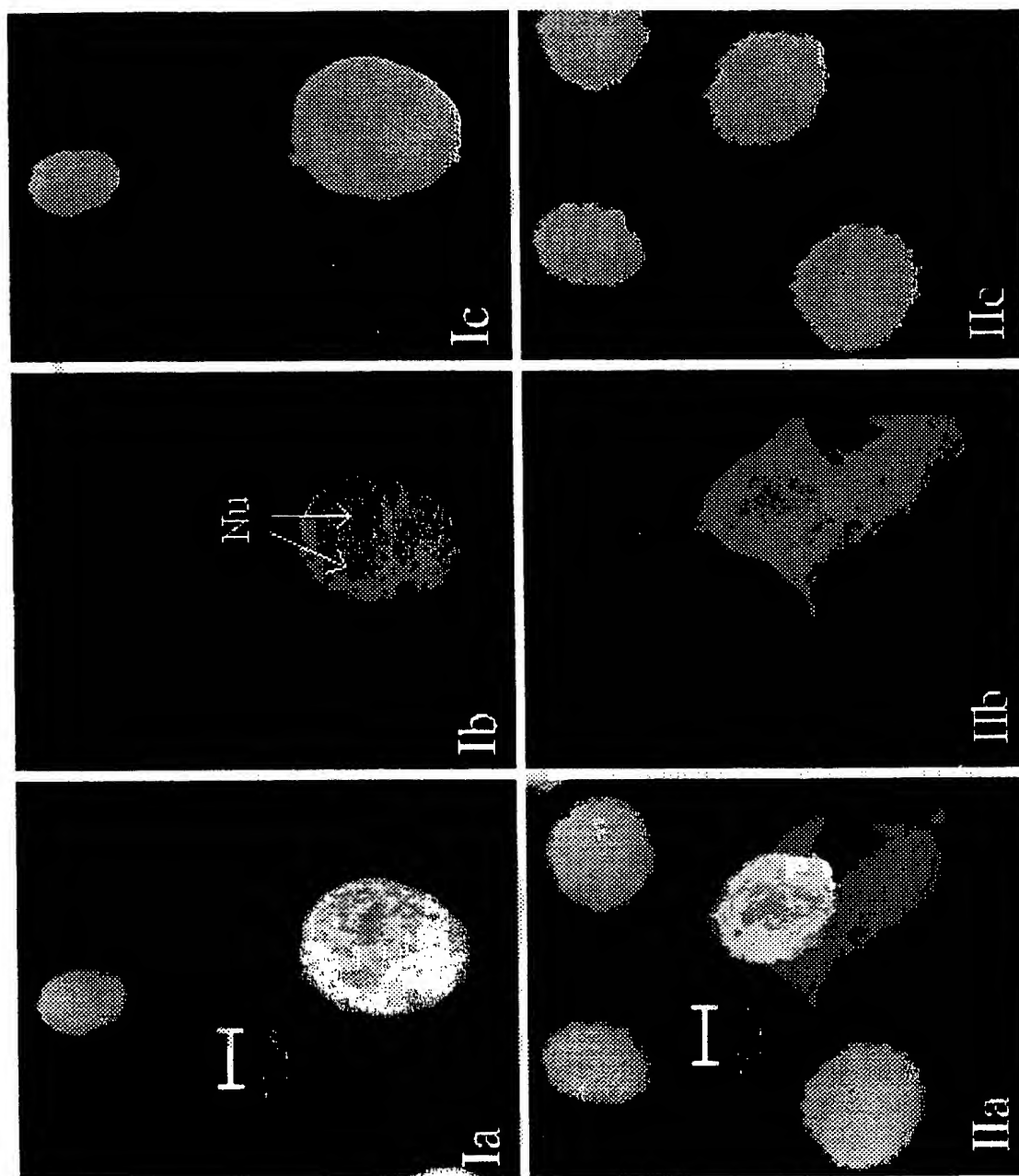
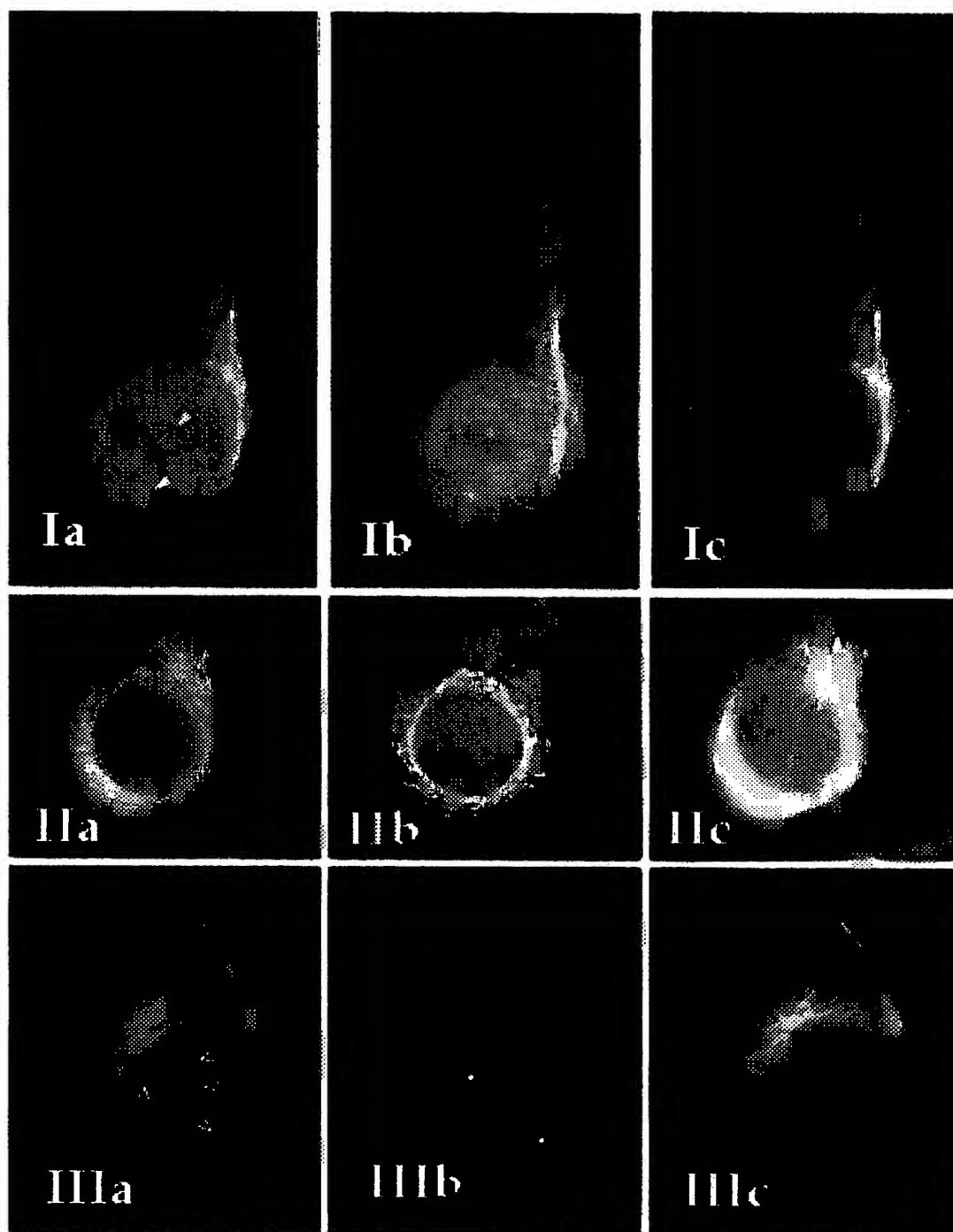
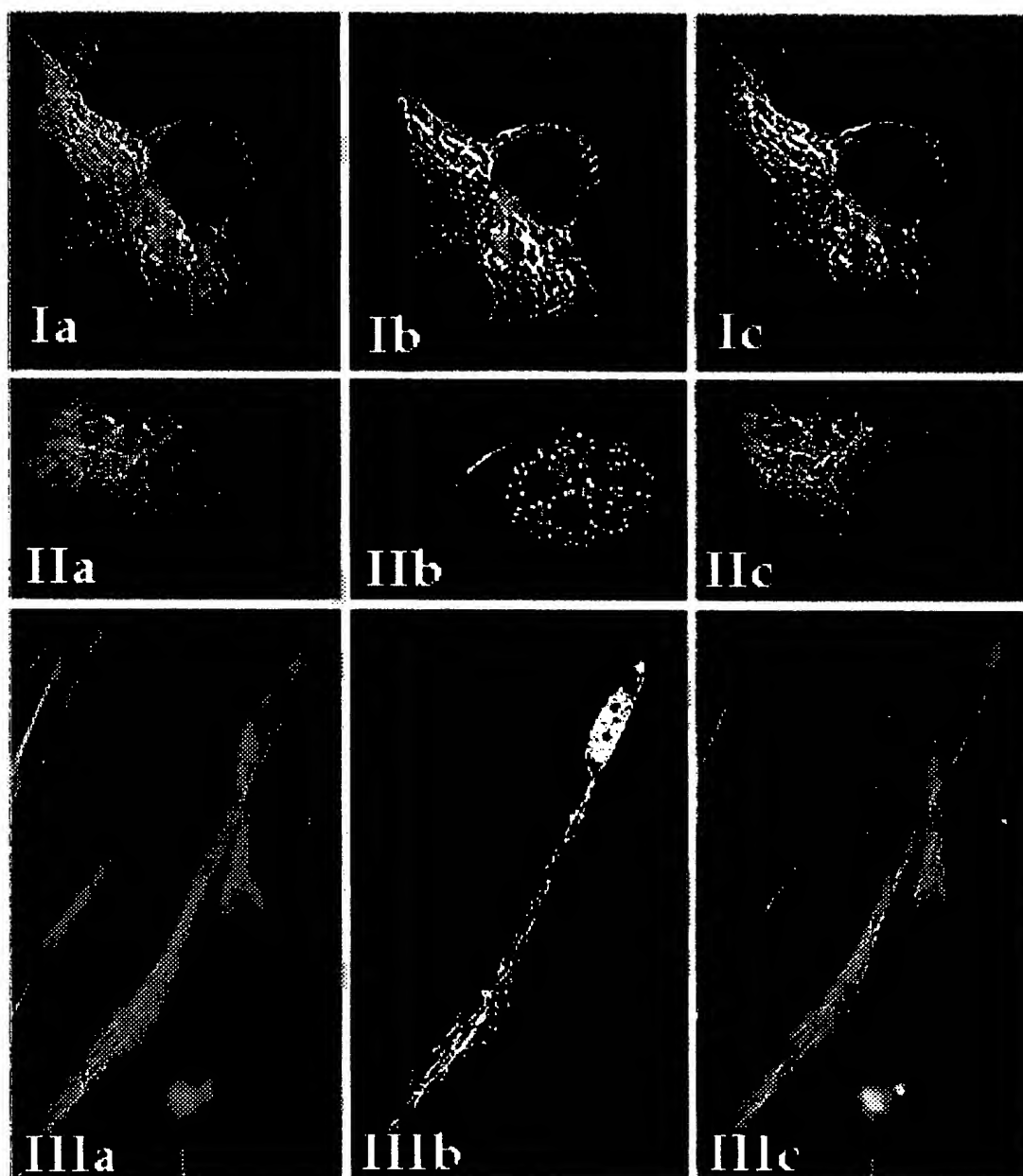


Figure 6

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**Figure 7**



**Figure 8**

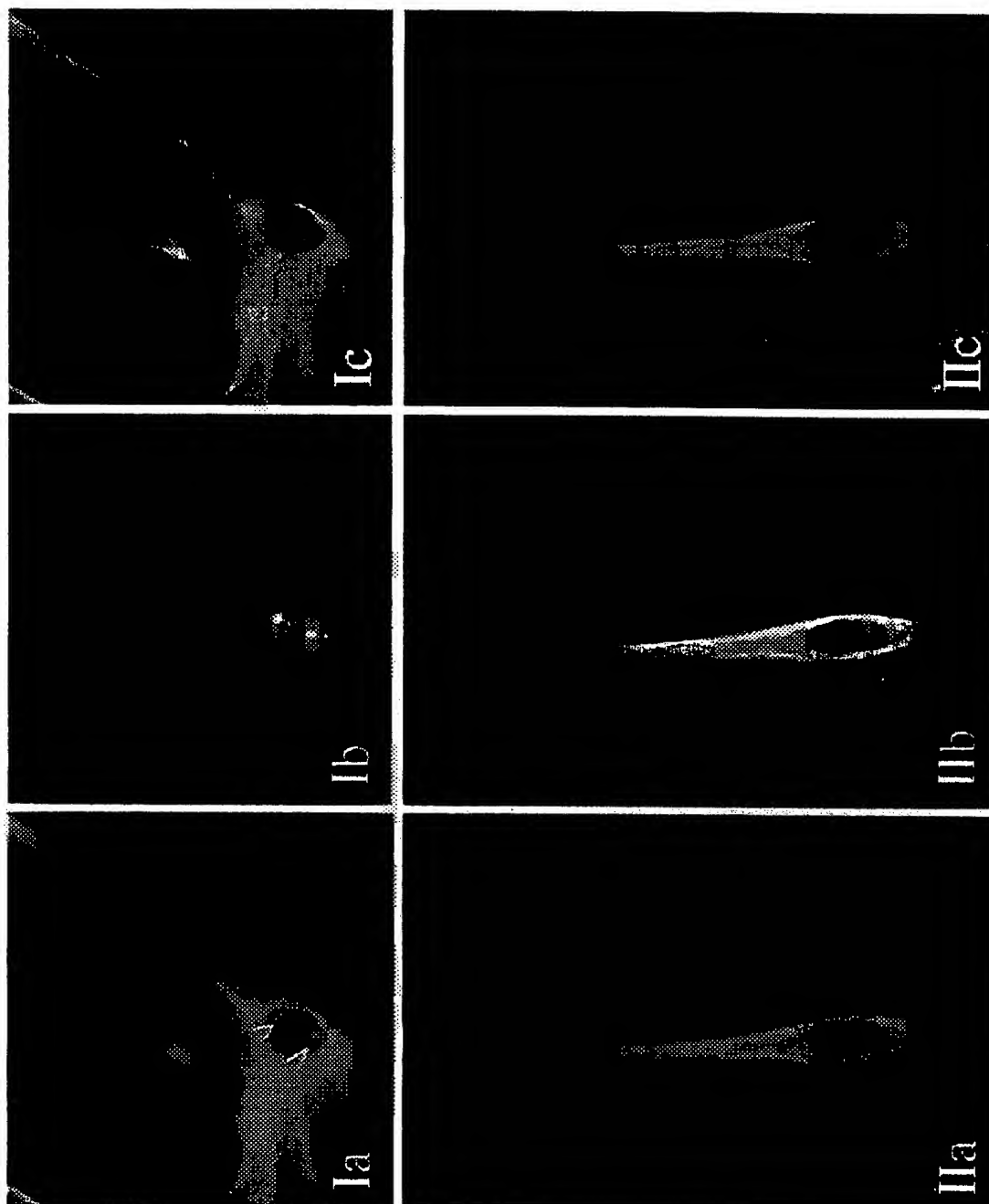
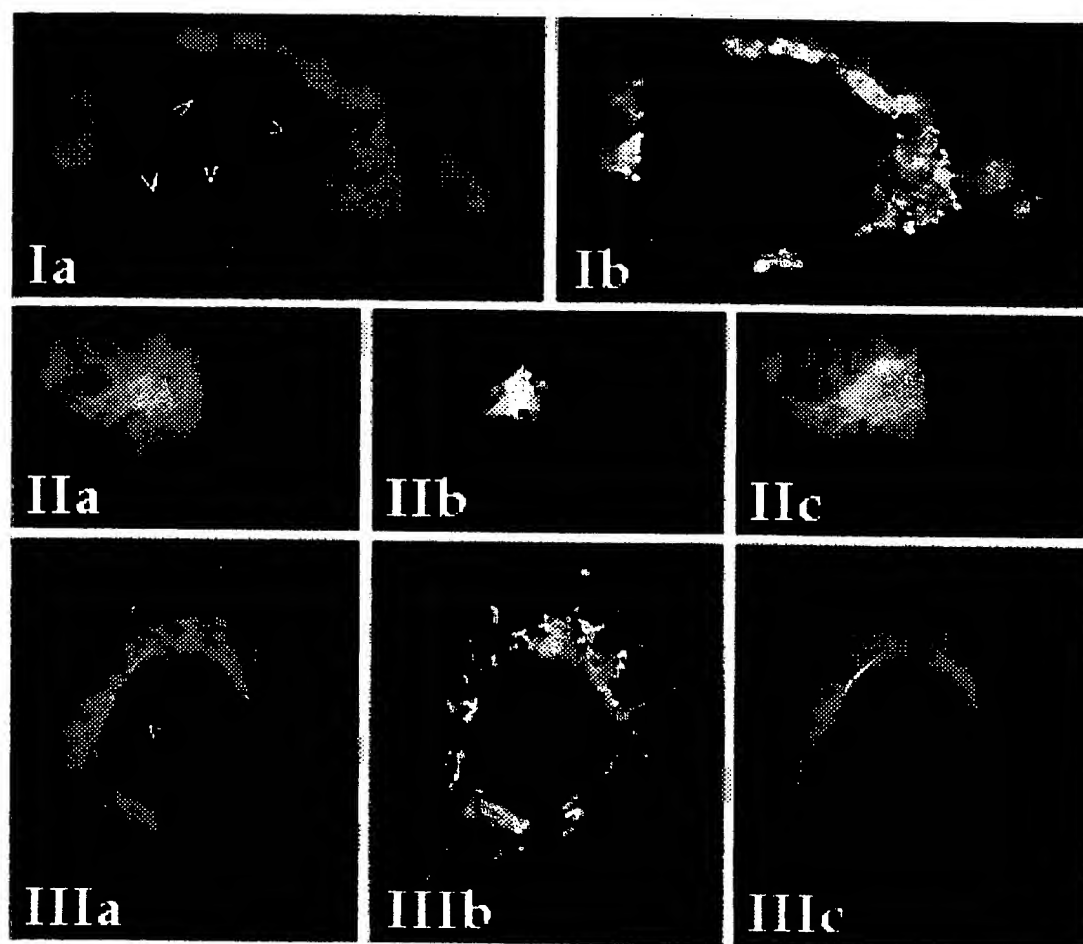
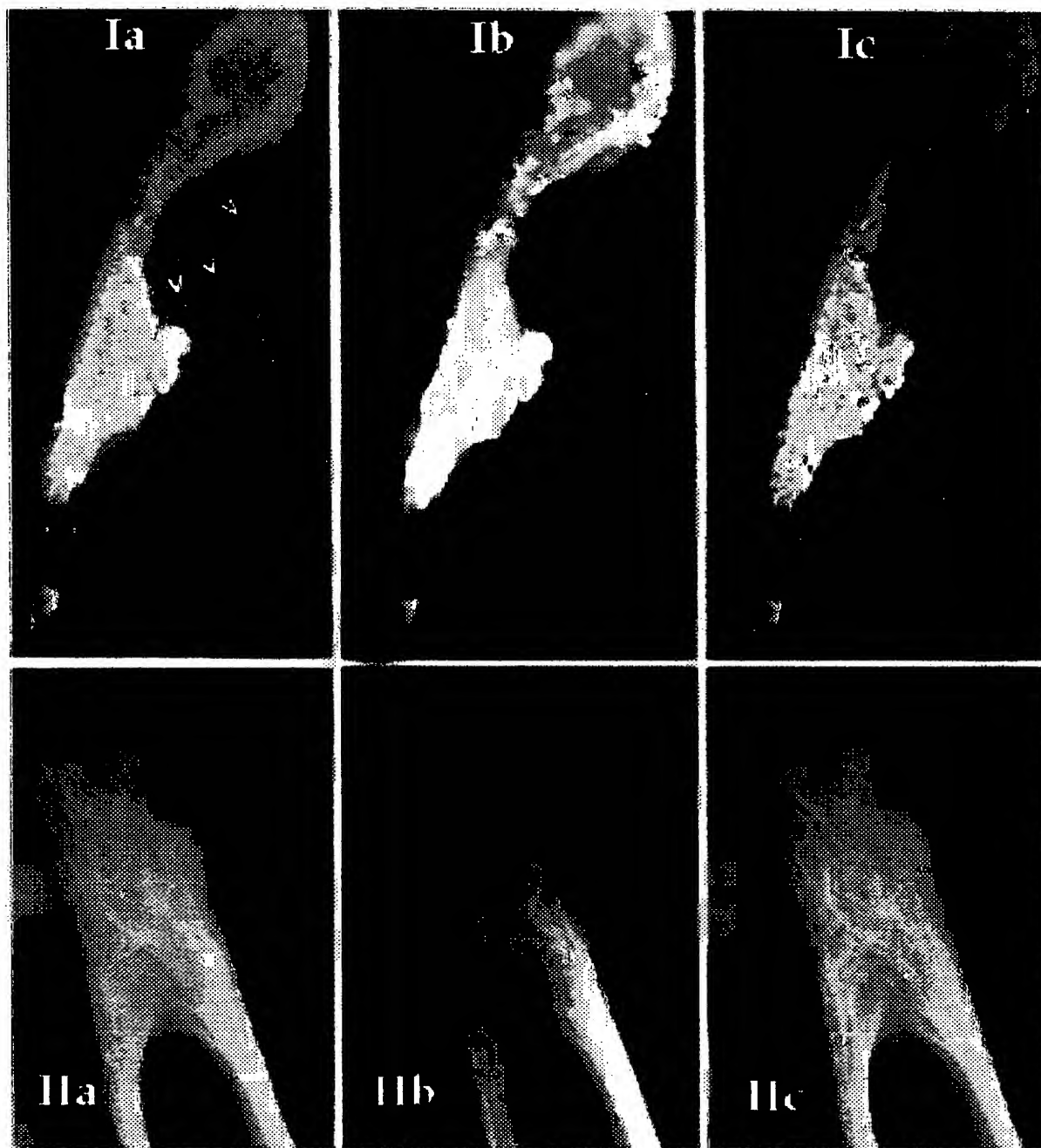


Figure 9

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**Figure 10**



**Figure 11**

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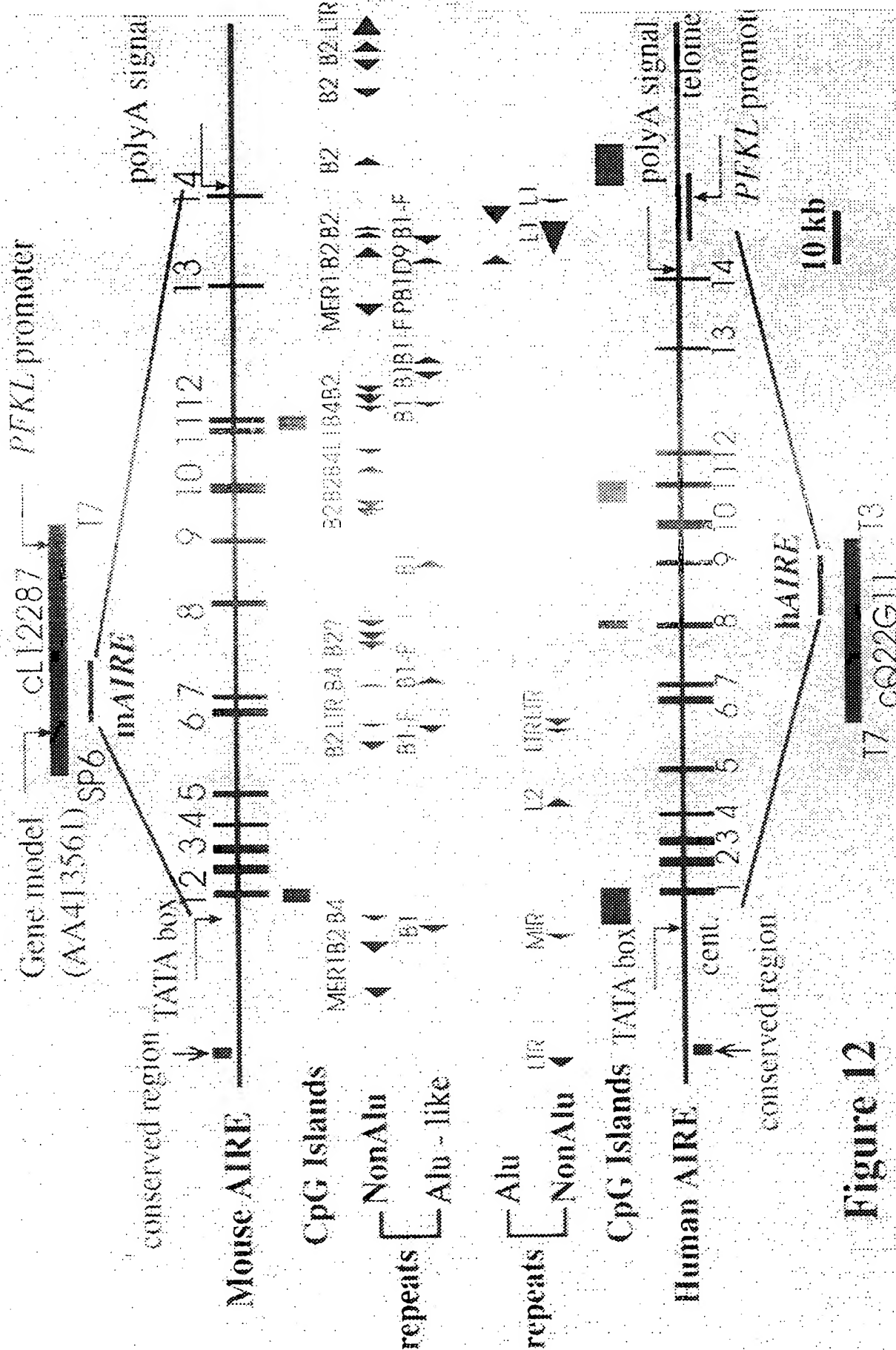


Figure 12



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HSAJ9610 (horizontal) vs. AF073797 (vertical)

09509595-070500

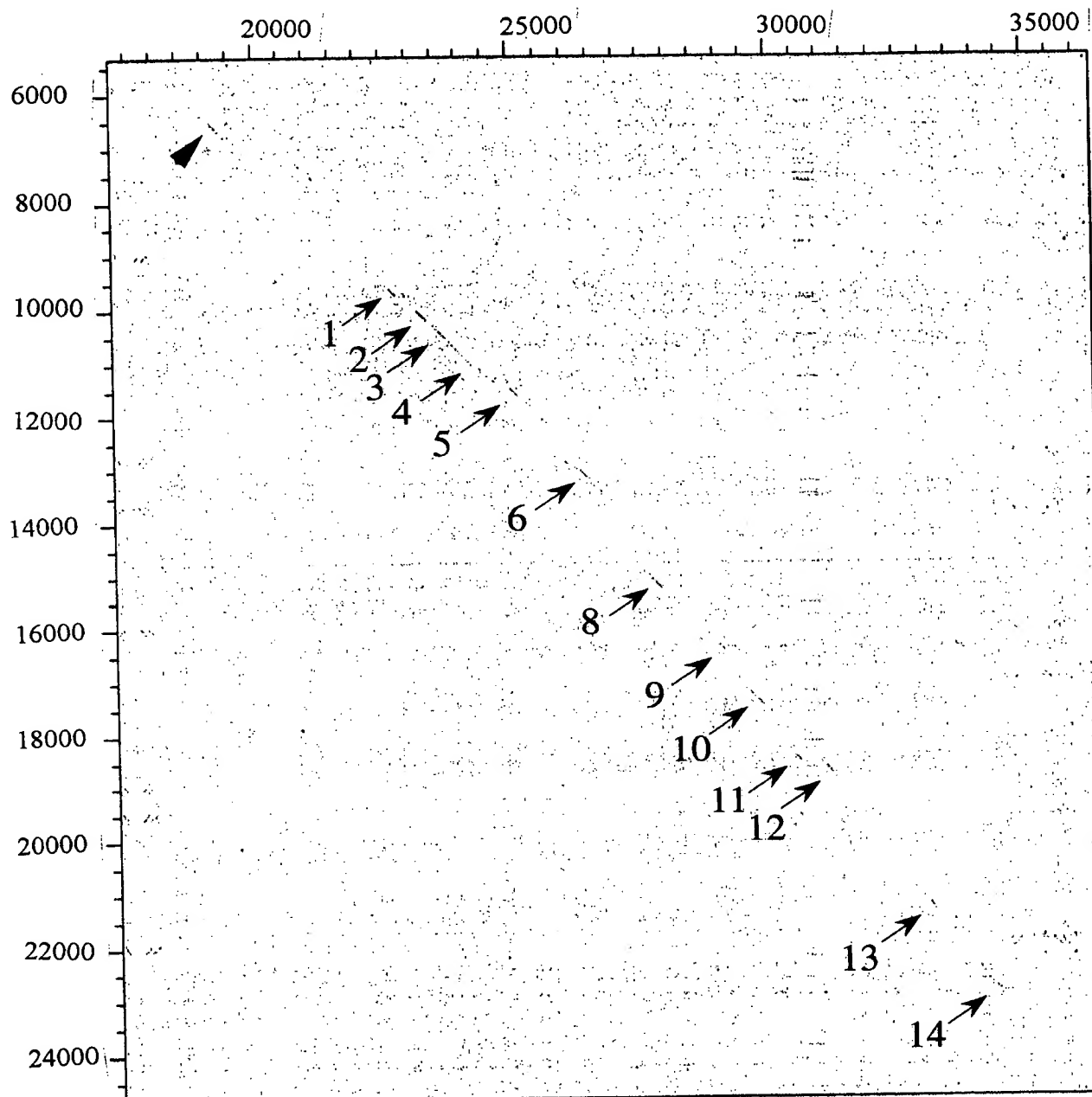


Figure 13A

0050/0" 56560560

6486 6575  
LAIRE GTGTGGACTG TCACGGAAAC CCCACGCTGT GATGGAAAGT CCAAAATTCT ACAGGAGTCT TTCTGTTGAT CTCCAGTCAG AGGCTGGGG  
LAIRE AAGGGGCTGG TGTGGAAAGC CCCACGGCAT GGTGGAAAGT CCGAAATTCT ACAGGGGCTT CTTGTTAAA CCTCCATGCA AGAGGCTGG  
19186 19275  
ons. --G-GG---G T---G-AA-C CCC---G---T G-TGGAAAGT CC-AAATTCT ACAGG-G-CT -T-TGTT-A- C--C--T--- AG-----GGG

Figure 13B

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	10	30	50	
1	ATGGCAGGTGGGGATGGAATGCTACGCCGTCTGCTGAGGCTGCACCGCACCGAGATCGCG			60
1	M A G G D G M L R R L L R L H R T E I A			20
	70	90	110	
61	GTGGCCATAGACAGTGCCTTTCCGCTGCTGCATGCTCTAGCCGACCACGACGTGGTCCCT			120
21	V A I D S A F P L L H A L A D H D V V P			40
	130	150	170	
121	GAGGACAAGTTCCAGGAGACGCTCCGTCTGAAGGAGAAGGAAGGCTGCCCCAGGCCTTC			180
41	E D K F Q E T L R L K E K E G C P Q A F			60
	190	210	230	
181	CACGCCCTGCTGTCCTGGCTCCTGACCCGGGACAGTGGGGCCATCCTGGATTTCTGGAGG			240
61	H A L L S W L L T R D S G A I L D F W R			80
	250	270	290	
241	ATTCTCTTTAAGGACTACAATCTGGAGCGGTACAGCCGCCTGCATAGCATCCTGGACGGC			300
81	I L F K D Y N L E R Y S R L H S I L D G			100
	310	330	350	
301	TTCCCAAAGATGTGGACCTAAACCAGTCCCGGAAAGGGAGAAAGCCCCCTTGCTGGTCCC			360
101	F P K D V D L N Q S R K G R K P L A G P			120
	370	390	410	
361	AAGGCCGCGGTACTGCCACCCAGACCCCCACCAAGAGAAAAGCACTGGAGGAGCCTCGA			420
121	K A A V L P P R P P T K R K A L E E P R			140
	430	450	470	
421	GCCACCCCAACAGCAACTCTGGCCTCAAAGAGCGTCTCCAGCCCAGGCTCCACCTGAAG			480
141	A T P P A T L A S K S V S S P G S H L K			160
	490	510	530	
481	ACTAAGCCCCCTAAGAAGCCAGATGGCAACTTGGAGTCAACAGCACCTTCCTCTTGGAAC			540
161	T K P P K K P D G N L E S Q H L P L G N			180
	550	570	590	
541	GGAATTCAGACCATGGCAGCTTCTGTCCAGAGAGCTGTGACCGTGGCCTCTGGGGATGTT			600
181	G I Q T M A A S V Q R A V T V A S G D V			200
	610	630	650	
601	CCAGGAACCCGAGGGGCCGTGGAAGGGATCCTTATCCAGCAGGTGTTTGAGTCAGGAAGA			660
201	P G T R G A V E G I L I Q Q V F E S G R			220
	670	690	710	
661	TCCAAGAAGTGCATTTCAGGTTGGGGGAGAGTTTTATACACCCAACAAGTTCGAAGACCCC			720
221	S K K C I Q V G G E F Y T P N K F E D P			240

Figure 14A

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	730	750	770	
721	AGTGGCAATTTGAAGAACAAGGCCCGGAGTGGTAGCAGCCTAAAGCCAGTGGTCCGAGCC			780
241	S G N L K N K A R S G S S L K P V V R A			260
	790	810	830	
781	AAGGGAGCCCAGGTCACTATACCTGGTAGAGATGAGCAGAAAGTGGGCCAGCAGTGTGGG			840
261	K G A Q V T I P G R D E Q K V G Q Q C G			280
	850	870	890	
841	G TTCCTCCCCTTCCATCCCTCCCCAGTGAGCCCCAGGTTAACCAGAAGAACGAGGATGAG			900
281	V P P L P S L P S E P Q V N Q K N E D E			300
	910	930	950	
901	TGTGCCGTGTGCCACGACGGAGGTGAGCTCATCTGTTGTGACGGCTGTCCCCGGGCCTTC			960
301	C A V C H D G G E L I C C D G C P R A F			320
	970	990	1010	
961	CACCTGGCTTGCTGTCCCCACCTCTGCAGGAGATCCCCAGTGGCCTCTGGAGATGCTCC			1020
321	H L A C L S P P L Q E I P S G L W R C S			340
	1030	1050	1070	
1021	TGCTGCCTCCAGGGCAGAGTCCAACAGAACCTGTCCCAGCCTGAGGTGTCCAGGCCCCCG			1080
341	C C L Q G R V Q Q N L S Q P E V S R P P			360
	1090	1110	1130	
1081	GAGCTACCTGCAGAGACCCCGATCCTCGTGGGACTGAGGTGAGCTTCAGAGAAAACCAGG			1140
361	E L P A E T P I L V G L R S A S E K T R			380
	1150	1170	1190	
1141	GGCCCATCCAGGGAGCTCAAAGCCAGCTCTGATGCTGCTGTACATATGTGAACCTGCTG			1200
381	G P S R E L K A S S D A A V T Y V N L L			400
	1210	1230	1250	
1201	GCCCCGCACCTGCAGCTCCTCTGCTGGAGCCTTCAGCACTGTGCCCTCTACTGAGTGCT			1260
401	A P H P A A P L L E P S A L C P L L S A			420
	1270	1290	1310	
1261	GGGAATGAGGGGCGGCCAGGTCCAGCACCAAGCGCGCATGCAGTGTGTGTGGCGATGGC			1320
421	G N E G R P G P A P S A R C S V C G D G			440
	1330	1350	1370	
1321	ACCGAGGTGTTGCGGTGTGCACACTGTGCCGCTGCCTTCCACTGGCGCTGCCACTTCCCG			1380
441	T E V L R C A H C A A A F H W R C H F P			460
	1390	1410	1430	
1381	ACGGCCGCCGCCGCGGGGACCAATCTCCGCTGCAAATCCTGCTCTGCAGACTCGACT			1440
461	T A A A R P G T N L R C K S C S A D S T			480
	1450	1470	1490	

Figure 14B

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1441	CCCACGCCAGGCACACCGGGCGAAGCTGTACCCACCTCTGGGCCCCGTCCAGCACCTGGG	1500
481	P T P G T P G E A V P T S G P R P A P G	500
	1510 1530 1550	
1501	CTTGCCAAGgtagGGGACGACTCTGCTAGTCACGACCCTGTTCTACATAGGGACGACCTG	1560
501	L A K V G D D S A S H D P V L H R D D L	520
	1570 1590 1610	
1561	GAGTCCCTCCTCAATGAGCACTCATTGACGGCATCCTGCAGTGGGCCATCCAGAGCATG	1620
521	E S L L N E H S F D G I L Q W A I Q S M	540
	1630 1650	
1621	TCACGCCCGCTGGCCGAGACACCACCCTTCTCTTCC	1656
541	S R P L A E T P P F S S	552

Figure 14C

M 1 2 3 4 5 6

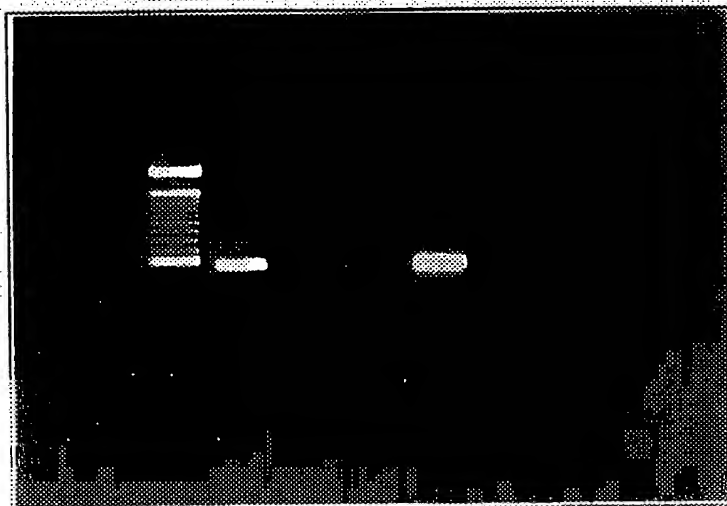


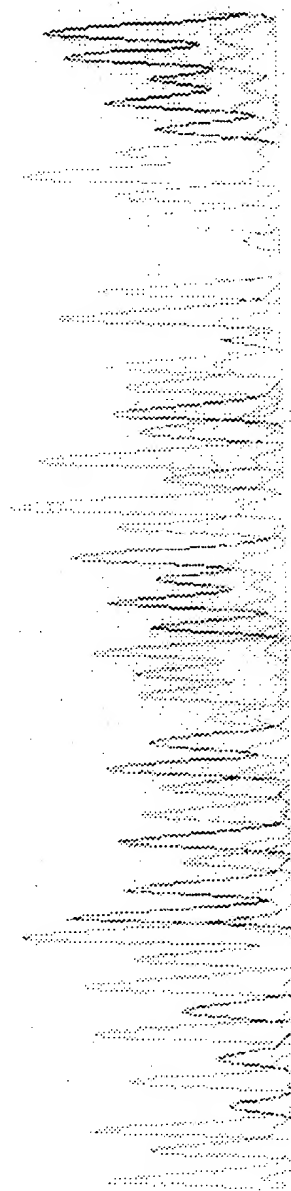
Figure 15

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Human AIRE	~MATDAALRR	LIRLHRTEIA	VAVDSAFPLL	HALADHDVVP	EDKFQETLHL
Mouse AIRE	MAGGDGMLRR	LIRLHRTEIA	VAIDSAFPLL	HALADHDVVP	EDKFQETLRL
Consensus	----D--LRR	LIRLHRTEIA	VA-DSAFPLL	HALADHDVVP	EDKFQETL-L
	51				100
Human AIRE	KEKEGCPQAF	HALLSWLLTQ	DSTAILDFWR	VLFKDYNLER	YGRLOPILDS
Mouse AIRE	KEKEGCPQAF	HALLSWLLTR	DSGAILDFWR	ILFKDYNLER	YSRLHSILDG
Consensus	KEKEGCPQAF	HALLSWLLT-	DS-AILDFWR	-LFKDYNLER	Y-RL--ILD-
	101				150
Human AIRE	FPKDVVLSQP	RKGRKPPAVP	KALVPPRPLP	TKRKASEEAR	AAAPAAALTPR
Mouse AIRE	FPKDVVLDNQ	RKGRKPLAGP	KAADVPPRPP	TKRKALEEPR	ATPPATLASK
Consensus	FPKDVVLD-Q-	<u>RKGRKP</u> -A-P	KA-V-PPR-P	<u>TKRKA</u> -EE-R	A--PA-L---
	151				200
Human AIRE	GTASPGSQLK	AKPPKKPES	AEQQLPLGN	GIQTMSASVQ	RAVAMSSGDV
Mouse AIRE	SVSSPGSHLK	TKPPKKPDGN	LESQHLPLGN	GIQTMAASVQ	RAVTVASGDV
Consensus	---SPGS-LK	-KPPKKP---	-E-Q-LPLGN	GIQTM-ASVQ	RAV---SGDV
	201				250
Human AIRE	PGARGAVEGI	LIQQVFESGG	SKKCIQVGGE	FYTPSKFED.	SGSGKNKARS
Mouse AIRE	PGTRGAVEGI	LIQQVFESGR	SKKCIQVGGE	FYTPNKFEDP	SGNLKNKARS
Consensus	PG-RGAVEGI	LIQQVFESG-	SKKCIQVGGE	FYTP-KFED-	SG--KNKARS
	251				300
Human AIRE	SSGPKPLVRA	KGAQGAAPGG	GEARLGQQGS	VPAPLALPSD	PQLHQKNEDE
Mouse AIRE	GSSLKPVVRA	KGAQVTIPGR	DEQKVGQQCG	VPPLPSLPSE	PQVNQKNEDE
Consensus	-S--KP-VRA	<u>KGAQ</u> ---PG-	-E---GQQ--	VP----LPS-	PQ--QKNEDE
	301				350
Human AIRE	CAVCRDGGEL	ICCDGCPRAF	HLACLSPPLR	EIPSGTWRC	SCLQATVQEV
Mouse AIRE	CAVCHDGGEL	ICCDGCPRAF	HLACLSPPLO	EIPSGLWRC	CCLQGRVQQN
Consensus	CAVC-DGGEL	ICCDGCPRAF	HLACLSPPL-	EIPSG-WRC	-CLQ--VQ--
	351				400
Human AIRE	QPRAEPRPQ	EPPVETPLPP	GLRSAGEEVR	GPPGEPLAGM	DTTLVYKHL
Mouse AIRE	LSQPEVSRPP	ELPAETPILV	GLRSASEKTR	GPSRELKASS	DAAVTYVNL
Consensus	----E--RP-	E-P-ETP---	GLRSA-E--R	GP--E--A--	D----Y--L-
	401				450
Human AIRE	APPSAAPLPG	LDSSALHPLL	CVGPEGQQNL	APGARCGVCG	DGTDVLRCTH
Mouse AIRE	APHPAAPL..	LEPSALCPLL	SAGNEGRPGP	APSARCSVCG	DGTEVLRCAH
Consensus	AP--AAPL--	L--SAL-PLL	--G-EG----	AP-ARC-VCG	DGT-VLRC-H
	451				500
Human AIRE	CAAAFHWRC	FPAGTSRPGT	GLRCRSCSGD	VTPAP.VEGV	LAP.SPAPLA
Mouse AIRE	CAAAFHWRC	FPTAAARPGT	NLRCKSCSAD	STPTPGTPGE	AVPTSGPRPA
Consensus	CAAAFHWRC	FP----RPGT	-LRC-SCS-D	-TP-P---G-	--P-S--R-A
	501				550
Human AIRE	PGPAK..DDT	ASHEPALHRD	DLESLLSEHT	FDGILQWAIQ	SMARPAAPFP
Mouse AIRE	PGLAKVGDD	ASHDPVLHRD	DLESLLNEHS	FDGILQWAIQ	SMSRPLAETP
Consensus	PG-AK--DD-	ASH-P-LHRD	DLESLL-EH-	FDGILQWAIQ	SM-RP-A--P
	551				
Human AIRE	S---				
Mouse AIRE	PFSS				
Consensus	----				

Figure 16

Exon 11  
 R A S P A P Q P T E A P L R P  
 3'-TGGGCGCTTGGTGGTGGGCGCGCGCGCGT-GCTGGGGG-5'  
 Δ Exon 10  
 Exon 9





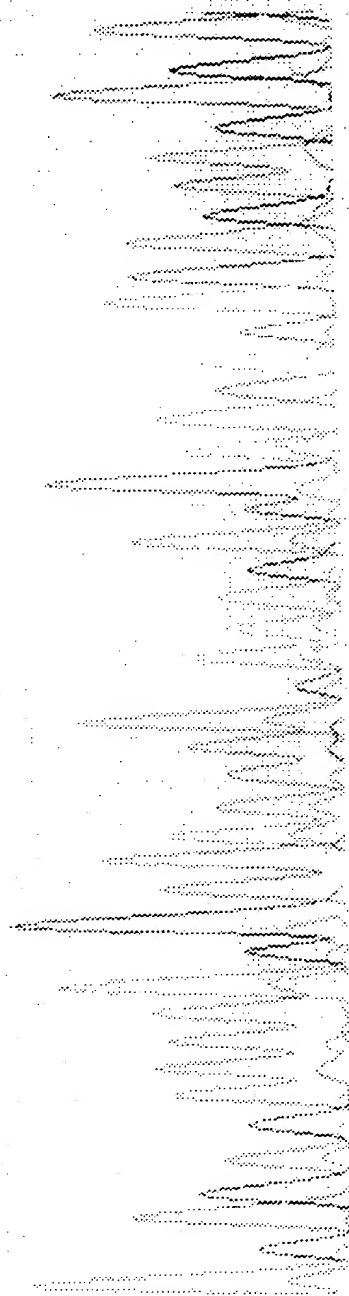
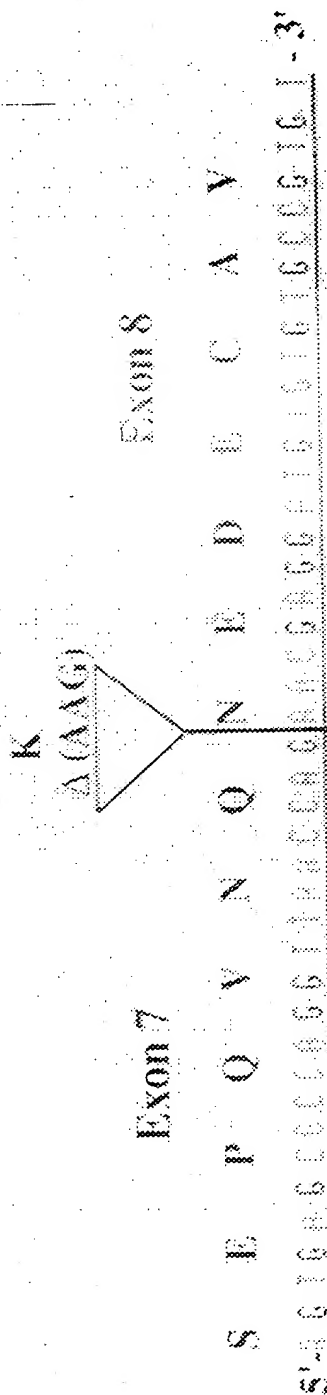


FIGURE 17B

005020" S6560560

P I T V  
Δ(TCCATATCACTG)

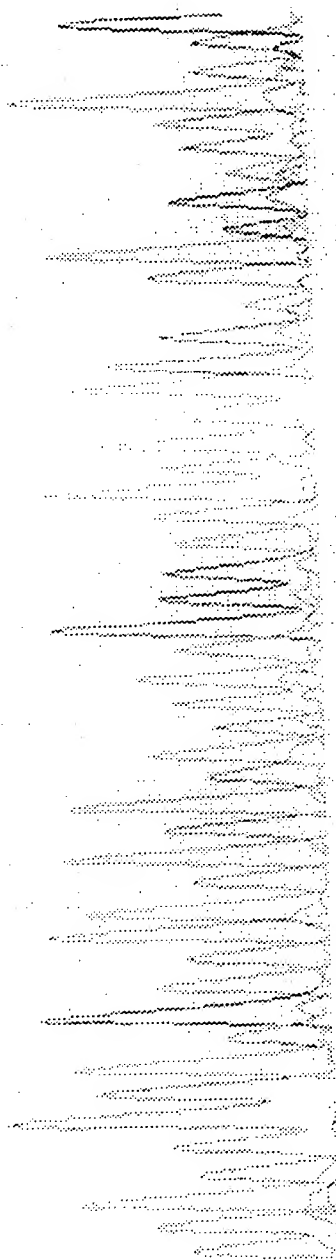
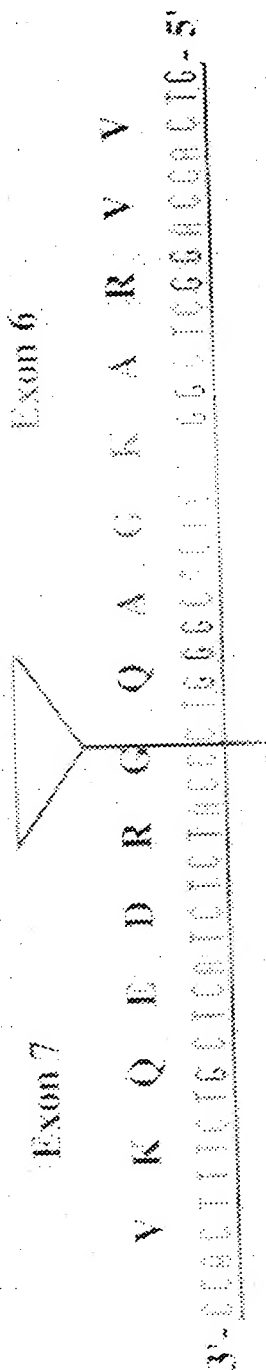


FIGURE 17C

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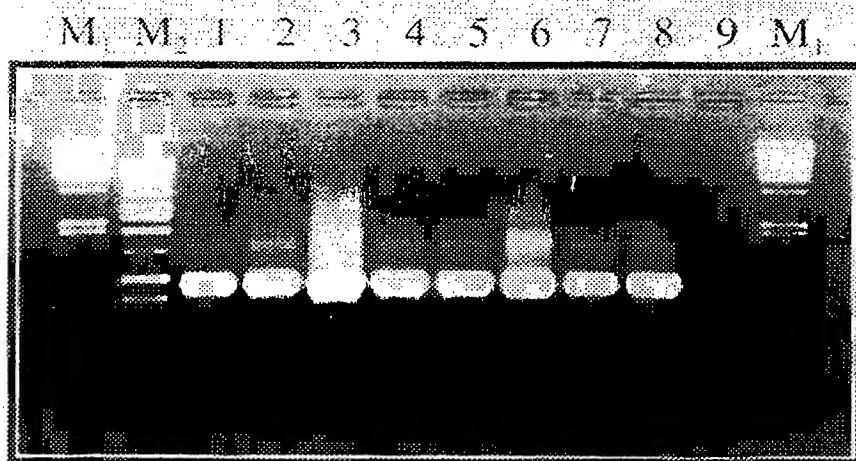


Figure 18